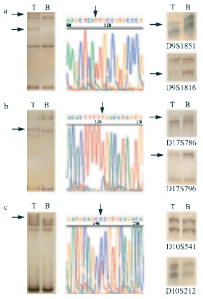
Research Snippets

Suggestive linkage of familial primary cutaneous amyloidosis to a locus on chromosome 1q23

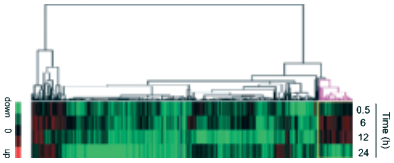
Familial primary cutaneous amyloidosis (FPCA) is a relatively uncommon skin disorder occurring with increased incidence in South America, South-east Asia and Taiwan. Reports of linkage to diseased-associated HLA antigens have been inconclusive. In this study, linkage analysis was used to look at chromosome 1 where the gene encoding serum amyloid P component is located. This study of FPCA in nine Chinese families suggests that a possible locus might exist on 1q23, but that other loci occur, confirming genetic heterogeneity for this disorder.
Lin M-W, Lee D-D, Lin C-H *et al*. Suggestive linkage of familial primary cutaneous amyloidosis to a locus on chromosome 1q23. *Br J Dermatol* 2005; **152**: 29–36.

Somatic mutations in the *PTCH*, *SMOH*, *SUFUH* and TP53 genes in sporadic basal cell carcinomas



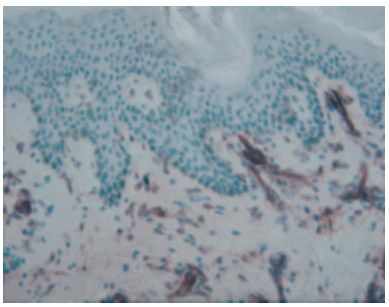
Basal cell carcinoma (BCC) of the skin is the most common human cancer. The genetic alterations underlying BCC development are only partly understood. Reifenberger *et al*. investigated 42 sporadic BCCs for mutations in 10 skin cancer-related genes (*PTCH*, *SMOH*, *SUFUH*, *GLI1*, *TP53*, *NRAS*, *KRAS*, *HRAS*, *BRAF* and *CTNNB1*). The results indicate that somatic mutations in the sonic hedgehog pathway genes *PTCH* and *SMOH*, as well as in the *TP53* tumor suppressor gene are important alterations in these tumors. Somatic mutations of *SUFUH* were restricted to individual BCCs. The other investigated genes could be excluded as important mutational targets.
Reifenberger J, Wolter M, Knobbe CB *et al*. Somatic mutations in the *PTCH*, *SMOH*, *SUFUH* and *TP53* genes in sporadic basal cell carcinomas. *Br J Dermatol* 2005; **152**: 43–51.

Analysis of genes responding to ultraviolet B irradiation of HaCaT keratinocytes using a cDNA microarray



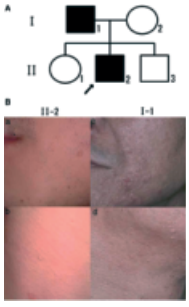
Ultraviolet B (UVB) irradiation causes many important biological changes in skin, which lead to pathophysiological alterations of the homeostatic environment. Lee *et al*. have performed cDNA microarray analysis to gain more insight into the molecular events provoked by UVB irradiation using HaCaT keratinocytes. Their analysis showed that the complexity of the transcriptional profile of the UVB response. Classification of these genes into nine functional categories revealed that UVB irradiation affected several biological processes. Their data provide a basis for the global characterization of UV-regulated gene expression.
Lee KM, Lee JG, Seo EY *et al*. Analysis of genes responding to ultraviolet B irradiation of HaCaT keratinocytes using a cDNA mocroarray. *Br J Dermatol* 2005; **152**: 52–59.

Angiogenesis in cutaneous photo-damage



UVB-irradiation of human skin resulted in pronounced dermal angiogenesis accompanied by the up-regulation of a potent angiogenesis factor, vascular endothelial growth factor (VEGF) and a down-regulation of an endogenous angiogenesis inhibitor, thrombospondin-1 (TSP-1). These newly formed blood vessels facilitated the infiltration of inflammatory cells into dermal tissue, resulting in the damage of dermal matrix components. Yano *et al* have previously reported that over-expression of TSP-1 in the skin prevented UVB-induced angiogenesis and cutaneous photo-damage. Inhibition of UV-induced angiogenesis may be a new thrapeutic strategy to prevent cutaneous photo-damage.
Yano K, Kadoya K, Kajiya K *et al*. Ultraviolet B irradiation of human skin induces an angiogenic switch that is mediated by upregulation of vascular endothelial growth and by downregulation of thrombospondin-1. *Br J Dermatol* 2005; **152**: 115–121.

Worldwide hot spot mutaion in Birt-Hogg-Dubé syndrome



Birt-Hogg-Dubé syndrome (BHD) is a rare genodermatosis characterized by multiple fibrofolliculomas, trichodiscomas, and acrochordons. Additional features include susceptibility to renal and colorectal carcinoma. Since the clinical feature include asymptomatic, small, soft, skin-colored papules, even an experienced clinical dermatologists may misdiagnose this condition as simply acrochordon. This study suggests that the hot spot mutation in the poly cytosine tract of exon 11 may have a worldwide distribution despite differing ethnic backgrounds. We strongly advise that physicians check for *BHD* hot spot mutations in any patients with wide spread acrochordon. Furthermore, physicians should suggest their patients to undergo investigation for kidney and colon lesions.
Kawasaki H, Sawamura D, Nakazawa H *et al*. Detection of 1733insC mutations in an Asian family with Birt-Hogg-Dube syndrome. *Br J Dermatol* 2005; **152**: 142–145.